IFNγR1 Deficiency Presenting with Visceral Leishmaniasis and Mycobacterium Avium infections mimicking HLH

Muhammad Khalid[1], Sonia Lemos[2], Katherine Myint-Hpu[1], Debbie Draper[1], Jennifer Stoddard[3], Julie Niemela[3], Stefania Pittaluga[4], Sergio Rosenzweig[3], Ottavia Delmonte[3], and Luigi D. Notarangelo[1]

[1]National Institute of Allergy and Infectious Diseases
[2]Centro Hospitalar e Universitario de Coimbra EPE
[3]National Institutes of Health
[4]National Cancer Institute

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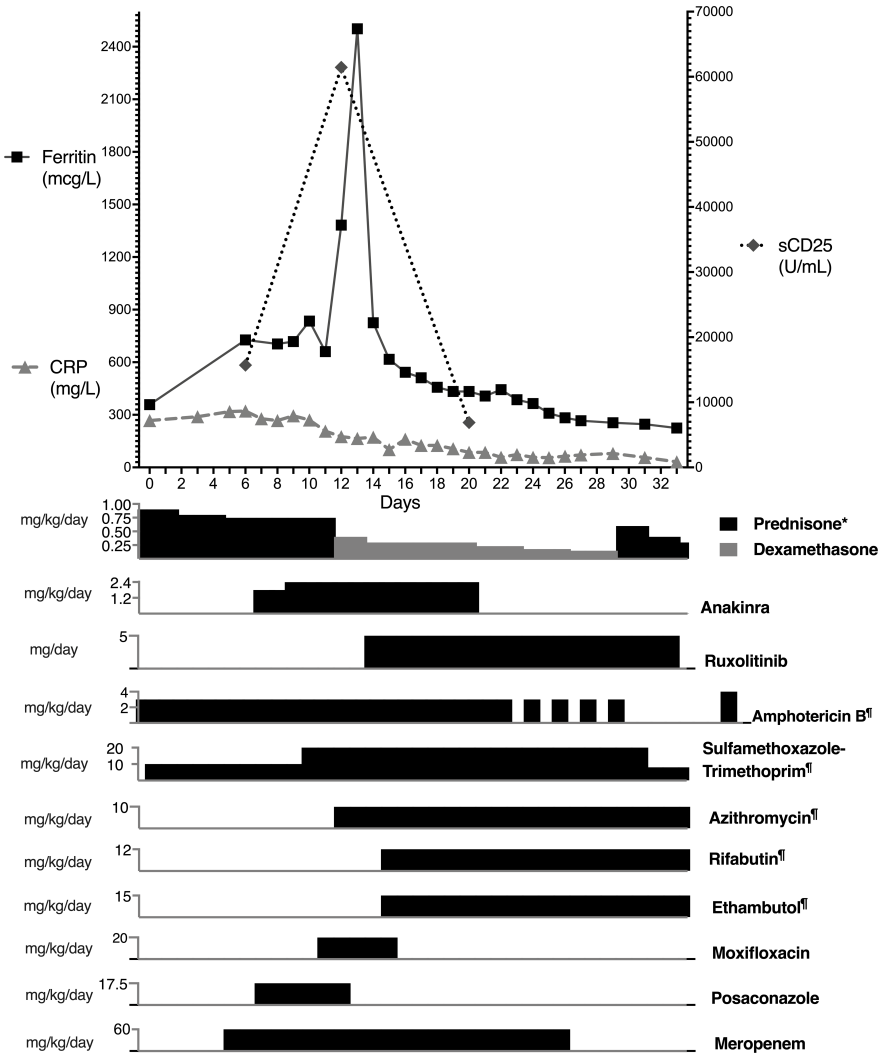

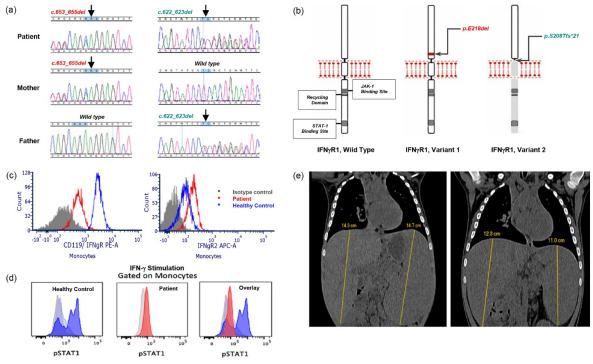

Figure 2. Demonstration of genetic, flow cytometric and radiographic changes in a patient with AR IFN-γR1 deficiency. (A) Sanger sequencing confirmation of *IFNGR1* variants in patient and parents, arrows indicating sites of mutation (B) Schematic illustration of the wild type IFNγR1 protein and the two variants; Variant 1: *p.E218del* and Variant 2: *p.S208Tfs*21* (C) Flow cytometric analysis in patient compared with healthy control, showing significantly reduced IFNγR1 expression and normal IFNγR2 expression (D) Flow cytometric analysis in patient compared with healthy control, showing absent STAT-1 phosphorylation in response to IFN-γ stimulation when gated on monocytes (E) Computed tomography images comparing hepatosplenomegaly at the peak of illness and at time of recovery.